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                E KAUFMAN STEPHEN?/AU
L1            20 S S E1 OR E2
L2        190114 S DYSTROPHY
L3           144 S L2 (S) (ALPHA (A) 7
L4             0 S L3 AND L1
L5             3 S L3 (S) SCAPULOPERONEAL
L6            63 DUP REM L3 (81 DUPLICATES REMOVED)
L7             4 S L6 (S) (ANTI OR ANTIBOD?)
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WEST Search History

DATE: Friday, June 20, 2003

Set Name side by side	Query	Hit Count	Set Name result set
DB=USPT,PGPB,JPAB,EPAB,DWPI; THES=ASSIGNEE; PLUR=YES; OP=ADJ			
L7	scapuloperoneal	6	L7
L6	l5 and l2	2	L6
L5	L3 same (alpha adj 7)	30	L5
L4	L3 same laminin	72	L4
L3	dystrophy	6292	L3
L2	kaufman-stephen-S.in.	33	L2
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L1	6207646.pn.	1	L1

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L7 ANSWER 1 OF 4 BIOTECHNO COPYRIGHT 2003 Elsevier Science B.V.
ACCESSION NUMBER: 2002:346634 BIOTECHNO
TITLE: Integrin .alpha.7.beta.1 in muscular dystrophy myopathy of unknown etiology
AUTHOR: Pegoraro E.; Cepollaro F.; Prandini P.; Marin A.; Panin M.; Trevisan C.P.; El-Messlemani A.H.; Tarone G.; Engvall E.; Hoffman E.P.; Angelini C.
CORPORATE SOURCE: Dr. E. Pegoraro, Neuromuscular Center, Department of Psychiatric Sciences, University of Padova, 35128 Padova, Italy.
E-mail: elena.pegoraro@unipd.it
SOURCE: American Journal of Pathology, (2002), 160(6) (2135-2143), 49 references
CODEN: AJPAA4 ISSN: 0002-9440
DOCUMENT TYPE: Journal; Article
COUNTRY: United States
LANGUAGE: English
SUMMARY LANGUAGE: English
AB To investigate the role of integrin .**alpha**.7 in muscle pathology, we used a "candidate gene" approach in a large cohort of muscular **dystrophy**/myopathy patients. **Antibodies** against the intracellular domain of the integrin .alpha.7A and .alpha.7B were used to stain muscle biopsies from 210 patients with muscular **dystrophy**/myopathy of unknown etiology. Levels of .alpha.7A and .alpha.7B integrin were found to be decreased in 35 of 210 patients (.apprx.17%). In six of these patients no integrin .alpha.7B was detected. Screening for .alpha.7B mutation in 30 of 35 patients detected only one integrin .**alpha**.7 missense mutation (the mutation on the second allele was not found) in a patient presenting with a congenital muscular **dystrophy**-like phenotype. No integrin .**alpha**.7 gene mutations were identified in all of the other patients showing integrin .**alpha**.7 deficiency. In the process of mutation analysis, we identified a novel integrin .**alpha**.7 isoform presenting 72-bp deletion. This isoform results from a partial deletion of exon 21 due to the use of a cryptic splice site generated by a G to A missense mutation at nucleotide position 2644 in integrin .**alpha**.7 cDNA. This spliced isoform is present in about 12% of the chromosomes studied. We conclude that secondary integrin .**alpha**.7 deficiency is rather common in muscular **dystrophy**/myopathy of unknown etiology, emphasizing the multiple mechanisms that may modulate integrin function and stability.
AB To investigate the role of integrin .**alpha**.7 in muscle pathology, we used a "candidate gene" approach in a large cohort of muscular **dystrophy**/myopathy patients. **Antibodies** against the intracellular domain of the integrin .alpha.7A and .alpha.7B were used to stain muscle biopsies from 210 patients with muscular **dystrophy**/myopathy of unknown etiology. Levels of .alpha.7A and .alpha.7B integrin were found to be decreased in 35 of 210 patients (.apprx.17%) . . . these patients no integrin .alpha.7B was detected. Screening for .alpha.7B mutation in 30 of 35 patients detected only one integrin .**alpha**.7 missense mutation (the mutation on the second allele was not found) in a patient presenting with a congenital muscular **dystrophy**-like phenotype. No integrin .**alpha**.7 gene mutations were identified in all of the other patients showing integrin .**alpha**.7 deficiency. In the process of mutation analysis, we identified a novel integrin .**alpha**.7 isoform presenting 72-bp deletion. This isoform results from a partial deletion of exon 21 due to the use of a cryptic splice site generated by a G to A missense mutation at nucleotide position 2644 in integrin .**alpha**.7 cDNA. This spliced isoform is present in about 12% of the chromosomes studied. We conclude that secondary integrin .**alpha**.7 deficiency is rather

common in muscular **dystrophy** myopathy of unknown etiology, emphasizing the multiple mechanisms that may modulate integrin function and stability.

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1: Muscular Dystrophies Links

A general term for a group of inherited disorders which are characterized by progressive degeneration of skeletal muscles (MUSCLE, SKELETAL).
Year introduced: 2000

Subheadings:

biosynthesis blood cerebrospinal fluid chemically induced classification complications congenital diagnosis diet therapy drug therapy economics embryology enzymology epidemiology ethnology etiology genetics history immunology metabolism microbiology mortality nursing parasitology pathology physiopathology prevention and control psychology radiography radionuclide imaging radiotherapy rehabilitation surgery therapy transmission ultrasonography ultrastructure urine

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Entry Terms:

- Muscular Dystrophy
- Dystrophies, Muscular
- Dystrophy, Muscular
- Myodystrophy
- Myodystrophies
- Myodystrophica
- Myodystrophicas
- Distal Myopathies
- Distal Myopathy
- Myopathies, Distal
- Myopathy, Distal
- Muscular Dystrophy, Limb-Girdle
- Dystrophies, Limb-Girdle Muscular
- Dystrophy, Limb-Girdle Muscular
- Limb-Girdle Muscular Dystrophies

- Limb-Girdle Muscular Dystrophy
- Muscular Dystrophies, Limb-Girdle
- Muscular Dystrophy, Limb Girdle
- Muscular Dystrophy, Scapuloperoneal
- Dystrophies, Scapuloperoneal Muscular
- Dystrophy, Scapuloperoneal Muscular
- Muscular Dystrophies, Scapuloperoneal
- Scapuloperoneal Muscular Dystrophies
- Scapuloperoneal Muscular Dystrophy

See Also:
- Mice, Inbred mdx

All MeSH Categories
Diseases Category
Musculoskeletal Diseases
Muscular Diseases
Muscular Disorders, Atrophic
Muscular Dystrophies
Glycogen Storage Disease Type VII
Muscular Dystrophy, Duchenne
Muscular Dystrophy, Emery-Dreifuss
Muscular Dystrophy, Facioscapulohumeral
Muscular Dystrophy, Oculopharyngeal
Myotonic Dystrophy

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Heredodegenerative Disorders, Nervous System
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Muscular Dystrophy, Duchenne
Muscular Dystrophy, Emery-Dreifuss
Muscular Dystrophy, Facioscapulohumeral
Muscular Dystrophy, Oculopharyngeal

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Diseases Category
Nervous System Diseases
Neuromuscular Diseases
Muscular Diseases
Muscular Disorders, Atrophic
Muscular Dystrophies
Muscular Dystrophy, Duchenne
Muscular Dystrophy, Emery-Dreifuss
Muscular Dystrophy, Facioscapulohumeral
Muscular Dystrophy, Oculopharyngeal

Myotonic Dystrophy

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Genetic Diseases, Inborn
Heredodegenerative Disorders, Nervous System
Muscular Dystrophies
Muscular Dystrophy, Duchenne
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Muscular Dystrophy, Facioscapulohumeral
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email: Debbie Constable

The GAPS INDEX

to Information on the Internet about Genetic Disorders and Birth Defects

Genetic Information and Patient Services, Inc. (GAPS)

Scapuloperoneal Myopathy

also known as:

Myogenic Facio-Scapulo-Peroneal Syndrome
Scapuloperoneal Muscular Dystrophy
Scapuloperoneal Syndrome, Myopathic Type

(as defined by the
National Organization for Rare Disorders)

Scapuloperoneal myopathy is a rare genetic disorder characterized by weakness and wasting of certain muscles.

Symptoms are usually limited to the shoulder blade area (scapula) and the smaller of the two leg muscle groups below the knee (peroneal).

Facial muscles may be affected in a few cases.

The leg symptoms often appear before the shoulder muscles become weakened.

The rate of progression of the disorder varies from case to case.

This condition can also occur in combination with other disorders.

Scapuloperoneal myopathy is inherited as an autosomal dominant trait.

Find more information on the Internet with

Google

Select name of the disorder

Search

SUPPORT GROUPS and information sites:

National Institute of Arthritis and Musculoskeletal and Skin Diseases
1 AMS Circle
Bethesda MD 20892-3675 USA
301 496-8188
877 226-4267
e-mail: NAMSIC@mail.nih.gov
Home Page: http://www.nih.gov/niams/

Muscular Dystrophy Association
3300 E. Sunrise Dr
Tucson AZ 85718 USA
520 529-2000
800 572-1717
e-mail: mda@mdausa.org
Home Page: http://www.mdausa.org

Scapuloperoneal Disease Association
610 Navesink Avenue
Ocean Gate NJ 08740 USA
908 269=0357
e-mail: N/A
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